CTC1 gene

CST telomere replication complex component 1

Normal Function

The *CTC1* gene provides instructions for making a protein that plays an important role in structures known as telomeres, which are found at the ends of chromosomes. Telomeres are short, repetitive segments of DNA that help protect chromosomes from abnormally sticking together or breaking down (degrading). In most cells, telomeres become progressively shorter as the cell divides. After a certain number of cell divisions, the telomeres become so short that they trigger the cell to stop dividing or to self-destruct (undergo apoptosis).

The CTC1 protein works as part of a group of proteins known as the CST complex, which is involved in the maintenance of telomeres. This complex is part of the special machinery that some cells use to copy (replicate) telomeres so they do not become too short as cells divide. Studies suggest that the CTC1 protein may also have roles in DNA replication unrelated to telomeres, but these functions are not well understood.

Health Conditions Related to Genetic Changes

Coats plus syndrome

At least 20 mutations in the *CTC1* gene have been identified in people with Coats plus syndrome. This disorder is characterized by an eye condition called Coats disease plus abnormalities of the brain, bones, gastrointestinal system, and other parts of the body.

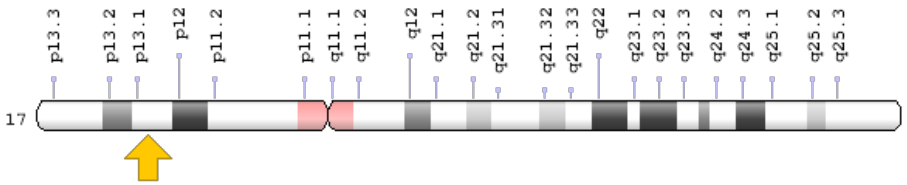
Most people with Coats plus syndrome have a mutation in one copy of the *CTC1* gene in each cell that eliminates the function of the CTC1 protein and a mutation in the other copy of the gene that reduces but does not eliminate the protein's function. This combination of mutations leaves only a small amount of functional CTC1 protein available to work as part of the CST complex. The resulting impairment of this complex affects the replication of telomeres, although the effect on telomere structure and function is unclear. Some studies have found that people with *CTC1* gene mutations have abnormally short telomeres, while other studies have found no change in telomere length. Researchers are working to determine how telomeres are different in people with *CTC1* gene mutations and how these changes could underlie the varied signs and symptoms of Coats plus syndrome.

dyskeratosis congenita

Chromosomal Location

Cytogenetic Location: 17p13.1, which is the short (p) arm of chromosome 17 at position 13.1

Molecular Location: base pairs 8,224,821 to 8,248,095 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AAF-132
- AAF132
- alpha accessory factor 132
- C17orf68
- conserved telomere capping protein 1
- CRMCC
- CST complex subunit CTC1
- CTS telomere maintenance complex component 1
- FLJ22170
- HBV DNAPTP1-transactivated protein B
- RP11-849F2.8
- tmp494178

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): Telomeres https://www.ncbi.nlm.nih.gov/books/NBK9863/#A634

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CTC1%5BTIAB%5D%29+AND+%28telomere*%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- CONSERVED TELOMERE MAINTENANCE COMPONENT 1
  http://omim.org/entry/613129

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CTC1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CTC1%5Bgene%5D
- HGNC Gene Family: CST complex
  http://www.genenames.org/cgi-bin/genefamilies/set/1343
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=26169
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/80169
- UniProt
  http://www.uniprot.org/uniprot/Q2NKJ3

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